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1: Neuron. 1992 Nov;9(5):815-30.

Transgenic mice with a rhodopsin mutation (Pro23His): a mouse model of autosomal dominant retinitis pigmentosa.

Olsson JE, Gordon JW, Pawlyk BS, Roof D, Hayes A, Molday RS, Mukai S, Cowley GS, Berson EL, Dryja TP.

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We inserted into the germline of mice either a mutant or wild-type allele from a patient with retinitis pigmentosa and a missense mutation (P23H) in the rhodopsin gene. All three lines of transgenic mice with the mutant allele developed photoreceptor degeneration; the one with the least severe retinal photoreceptor degeneration had the lowest transgene expression, which was one-sixth the level of endogenous murine rod opsin. Of two lines of mice with the wild-type allele, one expressed approximately equal amounts of transgenic and murine opsin and maintained normal retinal function and structure. The other expressed approximately 5 times more transgenic than murine opsin and developed a retinal degeneration similar to that found in mice carrying a mutant allele, presumably due to the overexpression of this protein. Our findings help to establish the pathogenicity of mutant human P23H rod opsin and suggest that overexpression of wild-type human rod opsin leads to a remarkably similar photoreceptor degeneration.

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